

Thalassemia

Thalassemia is an inherited blood disorder where the body cannot make normal amounts of hemoglobin, the oxygen-carrying protein inside red blood cells. Because hemoglobin is defective or scarce, red blood cells die early, causing anemia that ranges from mild to life-threatening.

THE ONE-LINE TRUTH

Thalassemia is a genetic failure to make enough of one type of hemoglobin chain, causing red blood cells to break apart too early and leading to lifelong anemia.

The chain — how it unfolds

1. Genetic mutation reduces or eliminates alpha-globin or beta-globin chain production — why: Without balanced chains, hemoglobin cannot form properly
2. Unpaired globin chains precipitate inside red cell precursors in bone marrow — why: Excess chains form toxic clumps that damage developing red blood cells
3. Ineffective erythropoiesis: many red cell precursors die before maturing — why: Damaged cells never leave the marrow, causing chronic anemia despite marrow expansion
4. Defective red cells that do reach the bloodstream hemolyze early — why: Rigid, damaged membranes are recognized and destroyed by the spleen within days to weeks
5. Chronic severe anemia triggers massive EPO release and marrow expansion — why: Body tries to compensate but marrow output is ineffective, causing bone thinning, fractures, and facial deformities
6. Extramedullary hematopoiesis and massive an enlarged liver and spleen (hepatosplenomegaly) develop — why: Liver and spleen take over blood production and removal, enlarging painfully and compressing nearby organs
7. Iron overload accumulates from increased absorption and repeated transfusions — why: Excess iron deposits in heart, liver, and endocrine glands, causing cardiomyopathy (heart muscle disease), cirrhosis (liver scarring), diabetes, and hypogonadism (low sex hormones)
8. End-organ failure: heart failure, liver cirrhosis, endocrine collapse, and early death — why: Untreated or undertreated iron toxicity and chronic anemia damage vital organs irreversibly

Spot it — what you see, and why

- Pale skin and pale mucous membranes (pallor): Low hemoglobin (oxygen-carrying protein in red blood cells) from abnormal globin chains that break down quickly reduces oxygen delivery to tissues
- Extreme tiredness and weakness (fatigue): Fewer healthy red blood cells mean less oxygen reaches muscles and organs, so the body feels exhausted
- Shortness of breath with activity: Low red blood cell count reduces oxygen-carrying capacity, so the lungs work harder to get enough oxygen during movement
- Yellowing of skin and eyes (jaundice): Abnormal red blood cells break down early, releasing bilirubin (yellow pigment from broken hemoglobin) faster than the liver can clear it
- Enlarged spleen (splenomegaly): The spleen traps and destroys defective red blood cells, becoming swollen from overwork and storing extra blood cells
- Bone pain and facial changes (frontal bossing, prominent cheekbones): Bone marrow expands to make more red blood cells to compensate for anemia, pushing outward on skull and facial bones

Red flags — act now

- Severe chest pain or sudden shortness of breath may indicate heart failure or acute chest syndrome
- High fever in a patient after splenectomy requires immediate antibiotics for possible overwhelming sepsis
- Sudden severe abdominal pain with rapid drop in hemoglobin suggests splenic sequestration crisis or rupture
- New irregular heartbeat or fainting may signal iron overload cardiomyopathy with dangerous arrhythmias

Treat it — in this order

1. Regular red blood cell transfusions (every 2-4 weeks for beta-thalassemia major) → Replaces defective red cells with healthy donor cells, raises hemoglobin to near-normal (9-10.5 g/dL), suppresses ineffective bone marrow expansion, prevents bone deformities and growth delay
2. Iron chelation therapy (removing excess iron with medication) → Binds iron in the blood and tissues so kidneys or intestines can remove it, prevents or reverses damage to heart, liver, and endocrine glands from hemosiderosis

3. Folic acid supplementation (1 mg daily) → Supports red blood cell production because chronic red blood cell destruction (hemolysis) and bone marrow hyperactivity use up folate (vitamin needed for DNA synthesis) quickly
4. Splenectomy (surgical removal of the spleen) → Reduces destruction of red blood cells, lowers transfusion frequency, and relieves abdominal discomfort from massive an enlarged spleen (splenomegaly)
5. Hematopoietic stem cell transplant (bone marrow transplant) → Only curative treatment; replaces defective bone marrow with healthy donor marrow that makes normal red blood cells, eliminates need for transfusions and chelation

Your top priorities at the bedside

1. Monitor hemoglobin and hematocrit levels closely — why: Thalassemia causes chronic anemia (low red blood cells) due to defective hemoglobin production, requiring regular monitoring to guide transfusion decisions and prevent organ damage from low oxygen (hypoxia) (low oxygen).
2. Assess for signs of iron overload — why: Repeated blood transfusions and increased intestinal iron absorption lead to iron buildup in the heart, liver, and endocrine glands, causing organ failure if untreated.
3. Administer chelation therapy as prescribed — why: Chelators (medicines that bind iron) like deferasirox or deferoxamine remove excess iron from the body to prevent or reverse damage to vital organs.